What is claimed is:

1. An oligonucleotide comprising a sequence complementary to the coding or non-coding strand of the MCOLN1 gene wherein said sequence includes a sequence consisting essentially of: 5'-AGC GGG CCG GAC TCA-3' (SEQ ID NO. 1), 5'-TAA CCA CCA TCG GAT CAA TGT C-3' (SEQ ID NO. 2), 5'-CTT GCT CTG TTG CCC AGG CT -3'(SEQ ID NO. 3), or 5'-CTC ACC GTG CTG GAA GAC ACT -3' (SEQ ID NO. 4), or a complementary sequence thereof.

2. An oligonucleotide of claim 1 wherein said sequence is SEQ ID NO. 1, SEQ ID NO. 2, SEQ ID NO. 3; or SEQ ID NO. 4, or a complementary sequence thereof.

3. An oligonucleotide comprising a sequence complementary to the coding or non-coding strand of the MCOLN1 gene wherein said sequence includes a sequence consisting essentially of: 5'- TCTG CCC ACA GTA CCT -3' (SEQ ID NO: 5), 5'- CTGC CCA CGG TAC CT -3' (SEQ ID NO: 6), or 5'- AGACC CAG GCC CAC AT- 3' (SEQ ID NO: 7), or a complementary sequence thereof.

4. The oligonucleotide of claim 4 wherein said sequence is SEQ ID NO: 5, SEQ ID NO: 6, or SEQ ID NO: 7, or a complementary sequence thereof.

5. The oligonucleotide of claim 1 wherein the oligonucleotide is conjugated to a detectable label.

6. The oligonucleotide of claim 3 wherein the oligonucleotide is conjugated to a detectable label.

7. The oligonucleotide of claim 6 wherein the detectable label comprises a donor fluorophore and quencher moiety.

8. The oligonucleotide of claim 7 wherein the donor fluorophore is 2'-chloro-7'-phenyl-1,4-dichloro-6-carboxyfluorescein (VIC), 6-carboxyfluorescein (FAM) or tetrachloro-6-carboxyfluorescein (TET).

9. The oligonucleotide of claim 8 wherein the quencher moiety is tetra-methylcarboxyrhodamine (TAMRA) or 4-(4'-dimethylaminophenylazo)benzoic acid (DABCYL).

10. The oligonucleotide of claim 8 wherein the sequence is 5'- TCTG CCC ACA GTA CCT -3' (SEQ ID NO: 5) and the molecular beacon pair is VIC and a quencher moiety.

9. The oligonucleotide of claim 8 wherein the sequence is 5'- CTGC CCA CGG TAC CT -3' (SEQ ID NO: 6) and the donor fluorophore is FAM.

10. The oligonucleotide of claim 8 wherein the sequence is 5'- AGACC CAG GCC CAC AT- 3' (SEQ ID NO: 7) and the donor fluorophore is TET.

11. A method of determining the presence of a Mucolipidosis IV mutant sequence in a nucleic acid, comprising,

a) contacting the nucleic acid with:

i) a first oligonucleotide primer that comprises a sequence complementary to a 15-30 bp segment of DNA between positions 5124-5524 of the MCOLN1 gene;

ii) a second oligonucleotide primer that comprises a sequence complementary to a 15-30 bp segment of DNA between positions 5541-5491 of the MCOLN1 gene,

iii) an oligonucleotide probe that comprises a sequence complementary to a 13-30 bp segment of DNA that includes position 5534 between of the MCOLN1 gene, wherein said probe is labeled with a detectable label which comprises a donor fluorophore and a quencher moiety, wherein said quencher moiety is optionally an acceptor fluorophore; and

b) conducting amplification by temperature cycling and monitoring the accumulation of amplified nucleic in real time by detecting an increase in donor fluorophore fluorescence or a decrease in acceptor fluorophore fluorescence which indicates the presence of the Mucolipidosis IV mutant sequence in the nucleic acid.

12. The method of claim 11 wherein the first oligonucleotide primer comprises a sequence that consists essentially of 5'-AGC GGG CCG GAC TCA-3' (SEQ ID NO. 1).

13. The method of claim 11 wherein the second oligonucleotide primer comprises a sequence that consists essentially of 5'-TAA CCA CCA TCG GAT CAA TGT C-3' (SEQ ID NO. 2).

14. The method of claim 11 wherein the probe comprises a sequence that consists essentially of 5'- CTGC CCA CGG TAC CT -3' (SEQ ID NO: 6).

15. The method of claim 11 wherein the donor fluorophore is 2'-chloro-7'-phenyl-1,4-dichloro-6-carboxyfluorescein (VIC), 6-carboxyfluorescein (FAM) or tetrachloro-6-carboxyfluorescein (TET).

16. The method of claim 11 wherein amplification by temperature cycling is with a DNA polymerase with 5' exonuclease activity and wherein binding of the probe to amplified nucleic acid results in degradation of the probe during DNA synthesis.

17. A method of determining the presence of a Mucolipidosis IV mutant sequence in a nucleic acid, comprising,

a) contacting the nucleic acid with;

i) a first oligonucleotide primer that comprises a sequence complementary to a 15-30 bp segment of DNA between positions 100-500 of the MCOLN1 gene,

ii) a second oligonucleotide primer that comprises a sequence complementary to a 15-30 bp segment of DNA between positions 6956-7356 of the MCOLN1 gene,

iii) an oligonucleotide probe that comprises a sequence complementary to a 13-30 bp segment of a fragment that is amplified using the first and second primer, wherein said probe is labeled with a detectable label which comprises a donor fluorophore and a quencher moiety, wherein said quencher moiety is optionally an acceptor fluorophore;

b) conducting amplification by temperature cycling and monitoring the accumulation of amplified nucleic in real time by detecting an increase in donor fluorophore fluorescence or a decrease in acceptor fluorophore fluorescence which indicates the presence of the Mucolipidosis IV mutant sequence in the nucleic acid.

18. The method of claim 17 wherein the first oligonucleotide primer comprises a sequence that consists essentially of 5'-CTT GCT CTG TTG CCC AGG CT -3'(SEQ ID NO. 3).

19. The method of claim 17 wherein the second oligonucleotide primer comprises a sequence that consists essentially of and 5'-CTC ACC GTG CTG GAA GAC ACT -3' (SEQ ID NO. 4).

20. The method of claim 17 wherein the probe comprises a sequence that consists essentially of and the probe in ii) is 5'- AGACC CAG GCC CAC AT- 3' (SEQ ID NO: 7).

21. The method of claim 17 wherein the donor fluorophore is 2'-chloro-7'-phenyl-1,4-dichloro-6-carboxyfluorescein (VIC), 6-carboxyfluorescein (FAM) or tetrachloro-6-carboxyfluorescein (TET).

22. The method of claim 17 wherein amplification by temperature cycling is with a DNA polymerase with 5' exonuclease activity and wherein binding of the probe to amplified nucleic acid results in degradation of the probe during DNA synthesis.

23. A method of detecting the presence of one or two Mucolipidosis IV mutant sequences in a nucleic acid, comprising,

a) contacting the nucleic acid with;

i) a first oligonucleotide primer that comprises a sequence complementary to a 15-30 bp segment of DNA between positions 100-500 of the MCOLN1 gene,

ii) a second oligonucleotide primer that comprises a sequence complementary to a 15-30 bp segment of DNA between positions 6956-7356 of the MCOLN1 gene,

iii) a first oligonucleotide probe that comprises a sequence complementary to a 13-30 bp segment of a fragment that is amplified using the first and second oligonucleotide primers, wherein said probe is labeled with a first detectable label which comprises a donor fluorophore and a quencher moiety, wherein said quencher moiety is optionally an acceptor fluorophore;

iv) a third oligonucleotide primer that comprises a sequence complementary to a 15-30 bp segment of DNA between positions 100-500 of the MCOLN1 gene,

v) a fourth oligonucleotide primer that comprises a sequence complementary to a 15-30 bp segment of DNA between positions 6956-7356 of the MCOLN1 gene, and

vi) a second oligonucleotide probe that comprises a sequence complementary to a 13-30 bp segment of a fragment that is amplified using the third and fourth primers, wherein said probe wherein said probe is labeled with a second detectable label which comprises a donor fluorophore and a quencher moiety, wherein said quencher moiety is optionally an acceptor fluorophore, and wherein said second detectable label is distinguishable from said first detectable label;

b) conducting amplification by temperature cycling and monitoring the accumulation of amplified nucleic in real time by detecting an increase in donor fluorophore fluorescence or an increase or decrease in acceptor fluorophore fluorescence, which indicates the presence of one or both of the Mucolipidosis IV mutant sequences in the nucleic acid.

24. The method of claim 23 wherein the first oligonucleotide primer comprises a sequence that consists essentially of 5'-AGC GGG CCG GAC TCA-3' (SEQ ID NO. 1).

25. The method of claim 23 wherein the second oligonucleotide primer comprises a sequence that consists essentially of 5'-TAA CCA CCA TCG GAT CAA TGT C-3' (SEQ ID NO. 2).

26. The method of claim 23 wherein the first probe comprises a sequence that consists essentially of 5'- CTGC CCA CGG TAC CT -3' (SEQ ID NO: 6).

27. The method of claim 23 wherein the third oligonucleotide primer comprises a sequence that consists essentially of 5'-CTT GCT CTG TTG CCC AGG CT -3'(SEQ ID NO. 3).

28. The method of claim 23 wherein the fourth oligonucleotide primer comprises a sequence that consists essentially of 5'-CTC ACC GTG CTG GAA GAC ACT -3' (SEQ ID NO. 4).

28. The method of claim 23 wherein the second probe comprises a sequence that consists essentially of 5'- AGACC CAG GCC CAC AT- 3' (SEQ ID NO: 7).

30. The method of claim 23 wherein the first or second donor fluorophore is 2'-chloro-7'-phenyl-1,4-dichloro-6-carboxyfluorescein (VIC), 6-carboxyfluorescein (FAM) or tetrachloro-6-carboxyfluorescein (TET).

31. The method of claim 23 wherein amplification by temperature cycling is with a DNA polymerase with 5' exonuclease activity and wherein binding of the probe to amplified nucleic acid results in degradation of the probe during DNA synthesis.

32. The method of claim 23 wherein said nucleic acid containing sample is also contacted with a third oligonucleotide probe comprising a sequence consisting essentially of 5'-TCTG CCC ACA GTA CCT -3' (SEQ ID NO: 5) that hybridizes to a wildtype sequence, wherein said third probe is labeled with a detectable label comprising a donor fluorophore and a quencher moiety wherein said quencher moiety is optionally an acceptor fluorophore, and wherein said third detectable label is distinguishable from said first and second detectable labels.

33. A kit for amplifying sequences of Mucolipidosis IV sequences comprising one or more oligonucleotides of claim 1.

31. The kit of claim 32 further comprising one or more oligonucleotides of claim 3.